Michigan Department of Community Health
MDCH
Jennifer M. Granholm, Governor
Janet Olszewski, Director

What is Marfan Syndrome?

Marfan syndrome is a genetic condition that occurs in males and females of all racial and ethnic backgrounds. It affects the body's connective tissue and can cause heart, eye, and bone problems. The symptoms vary widely, ranging from mild to severe. Marfan syndrome occurs in about 1 in 5,000 to 1 in 10,000 people.

Michigan Resources & Support

Children's Special Health Care Services
Family Phone Line
Toll-free: 1-800-359-3722
E-mail: ppp@michigan.gov
www.michigan.gov/cshcs

***Early On*® Michigan**
Toll-free: 1-800-EARLY ON
www.1800.earlyon.org

Michigan Birth Defects Program
Nurse Follow-up Coordinator
Toll-free: 1-866-852-1247
E-mail: BDRfollowup@michigan.gov

Michigan Genetics Connection
www.migeneticsconnection.org

National Resources & Support

National Marfan Foundation
Toll-free: 1-800-862-7326
www.marfan.org

Family Village
www.familyvillage.wisc.edu

GeneReviews
www.geneclinics.org/profiles/marfan/

Genetics Home Reference
www.ghr.nlm.nih.gov/condition=marfan syndrome

KidsHealth.org
kidshealth.org/kid/health_problems/birth_defect/marfan.html

March of Dimes
www.marchofdimes.com
click on Birth Defects & Genetics

National Institute of Arthritis and Musculoskeletal and Skin Diseases
www.niams.nih.gov/hi/topics/marfan/marfan.htm

How may Marfan syndrome affect my child?

Learning: In general, Marfan syndrome does not affect learning or intelligence. Loose joints may affect large motor skills in some children.

Physical: People with Marfan syndrome are often thin and tall with long limbs relative to the body's torso. Bones and cartilage may be affected, leading to curvature of the spine (scoliosis). The shape of the breast bone may cause a protruding or sunken chest. The joints may be very loose and flexible. The jaw is often narrow with a high-arched palate that can lead to dental crowding.

Medical: Symptoms can range from mild to very severe. Nearsighted vision (myopia) is typical, and often the first symptom identified. A dislocated lens (ectopia lentis) is a common sign often detected only by a special eye exam (slit lamp). Cataracts, glaucoma and retinal detachment may also occur. Heart problems, such as a floppy valve (mitral valve prolapse) are common and can cause shortness of breath and fatigue. The aorta may be wider and more fragile than normal. If not detected, there is a risk of aortic rupture (aneurysm) with serious complications including death.

How does Marfan syndrome occur?

Marfan syndrome is caused by a change in a gene (mutation). A child with Marfan syndrome may be the first and only family member affected, or the gene may be passed down from an affected parent. Parents of a newly diagnosed child should be checked carefully to look for signs of Marfan syndrome. There is a 1 in 2 chance that each child will have Marfan syndrome when a parent carries the gene. Genetic counseling is recommended for parents to learn about possible risks for other family members.

How is Marfan syndrome treated?

Marfan syndrome cannot be cured, but many symptoms can be treated. It is important for a child with Marfan syndrome to be monitored by medical specialists who understand the condition. Most eye problems are corrected with glasses alone but sometimes other procedures are needed. Everyone with Marfan syndrome must be under the care of a heart specialist (cardiologist). Frequent monitoring by ultrasound (echocardiogram) is needed, and medication or surgical procedures may be required. Other therapies or treatments may be needed for health problems as they occur. Infants and toddlers (birth to 3 years) should be connected with *Early On*® Michigan if there are concerns about development; while children over 3 years of age should be referred for special education services if concerns arise. Children with Marfan syndrome and their families benefit from having a primary care physician who helps to coordinate their care with medical specialists and other community-based services.

For more information, call Michigan's Genetics & Birth Defects Program toll-free at 1-866-852-1247 or e-mail Genetics@michigan.gov

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